Joseph Lagas, Doctoral Candidate in Molecular Biology (Washington University in St. Louis), generously agreed to provide a layman's summary of the following Alport-centric research paper published in February 2022

NanoLuc reporters identify COL4A5 nonsense mutations susceptible to drug-induced stop codon readthrough

Omachi, Kohei et al.
***iScience*, February 7, 2022.**

There are many types of genetic mutations that cause Alport syndrome. One of these types is known as a nonsense mutation, which means that the collagen protein only gets partially made and cannot be used for its intended purpose. Recently, a new therapy called “readthrough” therapy has been introduced which allows certain nonsense mutations to be bypassed so the full protein can be made. Dr. Kohei Omachi in Dr. Miner’s group tested this readthrough therapy on nonsense mutations found in Alport syndrome in a petri dish and discovered that this therapy has the potential to be successful in treating several common nonsense mutations. The next steps will be to determine why certain nonsense mutations are resistant to this therapy and how to apply these potential treatments in animals and humans.